

A rare study - a survey of the general practitioner's perspective on the problem of rare diseases

Background. Survey studies performed by Rare Diseases Sweden and EURORDIS have shown that patients with rare diseases are dissatisfied with care in general, and primary care in particular, along with the level of knowledge about rare diseases. To complete the picture we performed a similar study from the physician's perspective.

Purpose. The purpose of the study was to identify, from a general physician's own perspective, any needs of i) quality improvement in the overall care of patients with rare diseases, ii) training and iii) information on rare diseases among general practitioners.

Method. An invitation to participate in a web-based questionnaire was sent out to 1133 of the Swedish specialists and resident physicians in general medicine. In total, 293 physicians, distributed evenly geographically across the country, responded. The questions were formulated to highlight the problems that patients with rare diseases face in the primary care from the professional perspective. The questions dealt with diagnosis and coordination of care, and the doctors' knowledge, education and sources of information on rare diseases.

Results. The particular patient group was perceived as problematic primarily because of its heterogeneity and the lack of guidelines for the treatment of rare diseases. Communication between general practitioners and other health care providers regarding this patient group was experienced as inadequate. Furthermore, the GPs found it difficult to coordinate all different individual needs these patients had in a satisfactory manner. However, in general the physicians were satisfied with the primary care provided for the patient group, and they had also the perception that the patients in general were satisfied with the primary care. 70% of the GPs reported that they had acquired most of their knowledge of rare diseases on their own. Over 70% indicated they would be helped in their daily work by better knowledge about sources of information on rare diseases. Existing international information sources, such as Orphanet (orpha.net) and Medscape (emedicine.medscape.com), were not well known among the Swedish GPs and only 59% used the official Swedish National Board of Health and Welfare website on rare diseases.

Summary and Conclusions. The study indicate that the primary health care of patients with rare diseases can be improved. The weaknesses in this health care sector were perceived to be mainly of organizational nature. Communication and coordination can be improved, and additional support for the GPs, such as treatment guidelines and recommendations for additional rare disorders, is suggested to provide the care these patients need. Some of the existing high quality sources of information on rare diseases are insufficiently known. Better dissemination of knowledge about them can be one way to improve the care for the affected patients and to improve the work situation for the clinicians.

- TITLES IN CAPITAL LETTERS, **Authors in bolds, Last name and Initials**, Institutions and e-mail in Arial normal size 11 letters. Provide text without free spaces, use international abbreviations or clarify them in the text and respect the space available in the form. The summary must clarify the objectives of the work, the used methods, the results and conclusions.
- The **deadline** for abstracts submission is **December 15th 2011**.